Crystal formation on the meibomian glands is a diagnostic sign for pseudohypoaldosteronism type I

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Introduction

pseudohypoaldosteronism type I (PHA1) is a rare disease of mineralocorticoid resistance (MR). Neonatal manifestation leads to life-threatening dehydration due to massive salt-loss, acidosis and, frequently, failure to thrive. Two clinically and genetically distinct forms exist, the systemic and renal PHA1 caused by mutations in the subunit genes *(SCNN1A*, *SCNN1B*, *SCNN1G)* of the epithelial sodium channel (ENaC) and the mineralocorticoid receptor coding gene *NR3C2 [1]*.

Case report

- Eutrophic term male neonate, Polyhydramnia, otherwise pregnancy uneventful, birth weight 3770 g (50-90 pc), height 57 cm (>90 pc), head circumference 37,5 cm (>90 pc)
- Family history inconspicuous, 2´nd child of non-related healthy parents of Turkish decent
- Within the first hours of life poor feeding and mild respiratory distress; suspecting neonatal infection antibiotic therapy and intravenous fluid support, which lead to apparent stabilisation.
- on the 6th day of life leading to progressive deterioration, the boy showed clinical signs of dehydration and respiratory insufficiency. Lab results (tab. 1) demonstrated severe electrolyte imbalance suggestive of congenital adrenal hyperplasia with hyponatraemia, hyperkalaemia and metabolic acidosis. However, a stress dose of hydrocortisone had no beneficial effect.

Results:

- Uexpectedly, normal 17-a-hydroxyprogesterone levels (17-a-OHP) in neonatal screening
- Elevated plasma renin and aldosterone concentrations at the same time
- Of note, impressive salt crystal formation on the eyelids macroscopically visible via the excretory meibomian glands (fig. 1) – a major clinical sign for severe manifestation of systemic PHA1
- Mutational analysis confirmed compound heterozygous *SCNN1B* mutations (fig. 2)

References:
1. *Bowden SA et al. Cases in Endocrinology 2013*
2. *Rajpoot SK et al. Endocrinology, Diabetes and Metabolism 2014*
3. *Riepe FG. Horm Res 2009*

Conclusions:

The clinical presentation of severe PHA1 includes salt crystal deposition on the eyelids and the epidermis (*miliaria crystallina*). Life-saving specific therapy depends on early clinical diagnosis [2, 3].

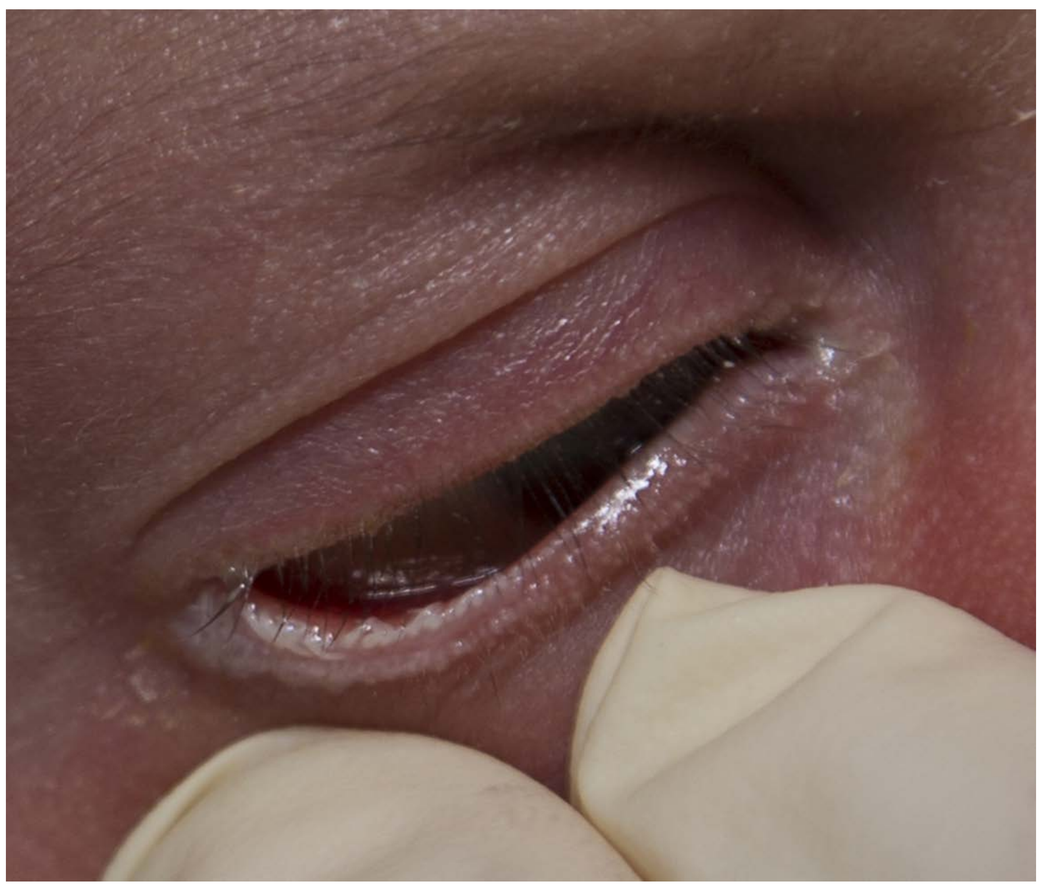

Fig. 1: Impressive salt crystal formation on the eyelids

Tab. 1: Biochemical data on day 6

Serum sodium (132-142 mmol/l)	125		Serum aldosterone (50-900 ng/l)	7368
Serum potassium (4-6,2 mmol/l)	8,08		plasma renin (2,6-27,7 ng/l)	633

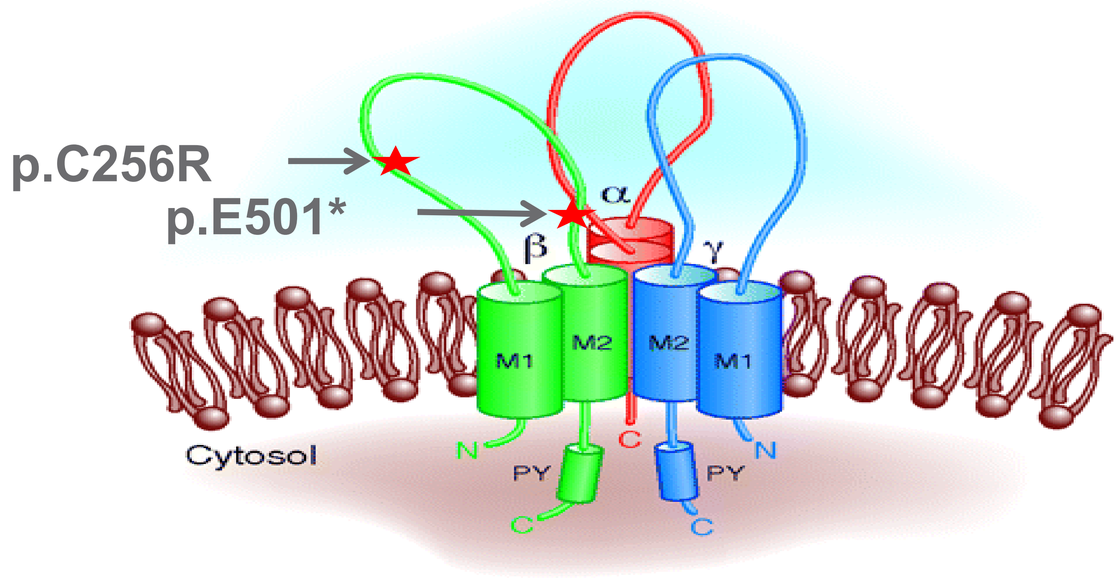

Fig. 2: SCNN1B:c.[766T>C];[1501G>T]

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